Class 19: RNASeq

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Summary of the Goal

One sample is obviously not enough to know what is happening in a population. You are interested in assessing genetic differences on a population scale. So, you processed about ~230 samples and did the normalization on a genome level. Now, you want to find whether there is any association of the 4 asthma-associated SNPs (rs8067378…) on ORMDL3 expression.

Q13. Examining the Dataset

> Q13: Read this file into R and determine the sample size for each genotype and their corresponding median expression levels for each of these genotypes.

```
#Read in the data and display the top to check
expr <- read.table("rs8067378_ENSG00000172057.6.txt")
head(expr)
```

```
##    sample geno      exp
## 1 HG00367  A/G 28.96038
## 2 NA20768  A/G 20.24449
## 3 HG00361  A/A 31.32628
## 4 HG00135  A/A 34.11169
## 5 NA18870  G/G 18.25141
## 6 NA11993  A/A 32.89721
```

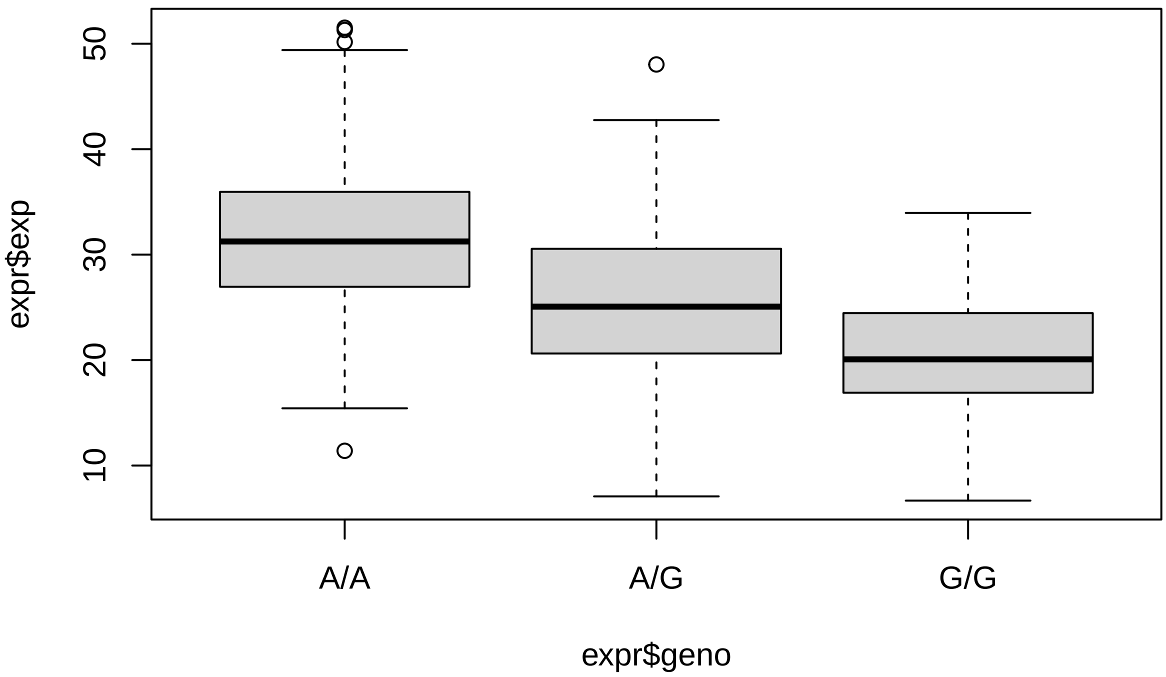
```
#Determine sample size total
nrow(expr)
```

```
## [1] 462
```

```
#Determine sample size for each genotype
table(expr$geno)
```

```
##
## A/A A/G G/G
## 108 233 121
```

```
#Generate a boxplot object grouped by genotype
base_box <- boxplot(expr$exp ~ expr$geno)
```



```
#Summarize the statisticsof eacg group
summary(base_box$stats)
```

```
##        V1              V2               V3
##  Min.   :15.43   Min.   : 7.075   Min.   : 6.675
##  1st Qu.:26.95   1st Qu.:20.626   1st Qu.:16.903
##  Median :31.25   Median :25.065   Median :20.074
##  Mean   :31.80   Mean   :25.215   Mean   :20.413
##  3rd Qu.:35.96   3rd Qu.:30.552   3rd Qu.:24.457
##  Max.   :49.40   Max.   :42.757   Max.   :33.956
```

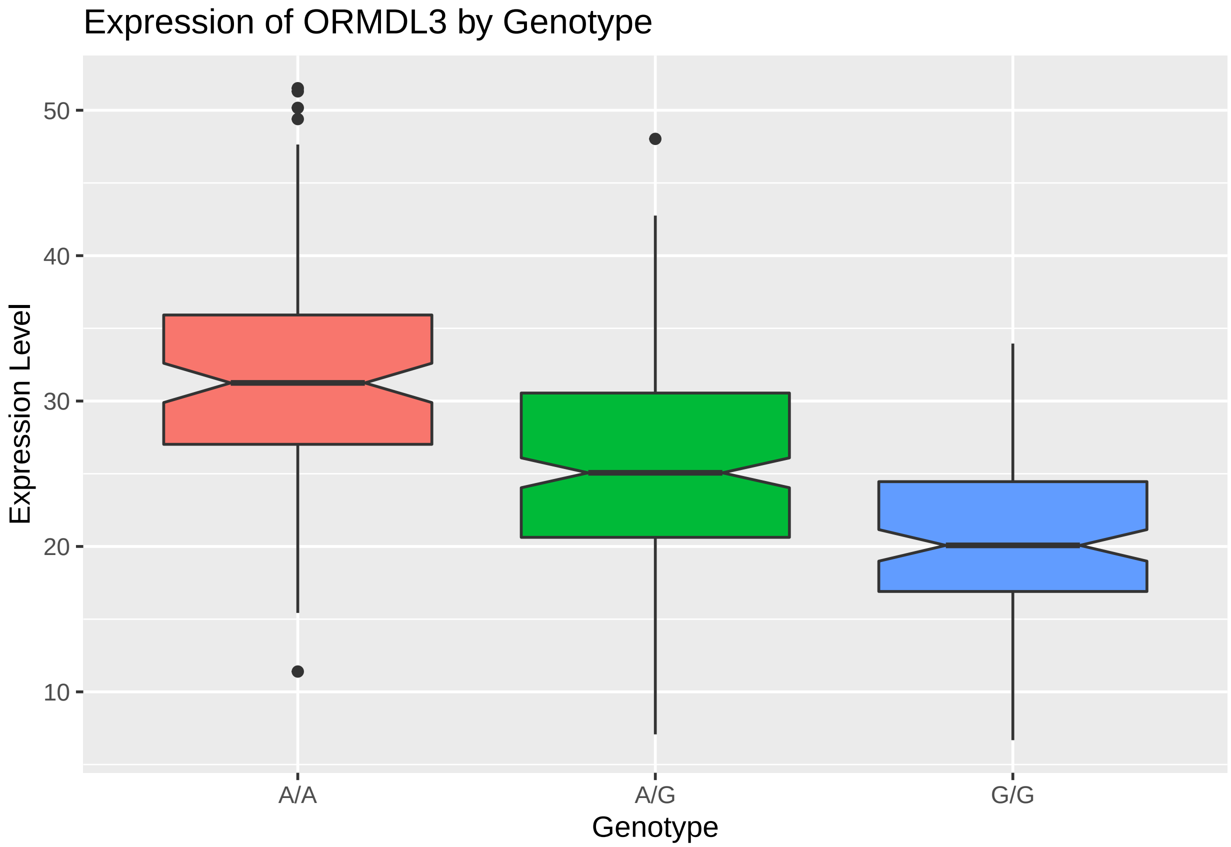
This shows the median expression levels for each genotype. For the A/A genotype it is 31.25, for the A/G genotype it is 25.065, and for the G/G genotype it is 20.074.

Q14. Using ggplot to Make a Cleaner Boxplot

```
library(ggplot2)
```

> Q14:Generate a boxplot with a box per genotype, what could you infer from the relative expression value between A/A and G/G displayed in this plot? Does the SNP effect the expression of ORMDL3?

```
#Build a better boxplot
ggplot(expr) + aes(geno, exp, fill=geno) +
  geom_boxplot(notch=TRUE, show.legend=FALSE) +
  xlab("Genotype") + ylab("Expression Level") + ggtitle("Expression of ORMDL3 by Genotype")
```



Based on this plot, it appears that the “G” variant SNP lowers the expression of ORMDL3.